

FIG. 1A

ESTs cluster 1  
ESTs cluster 2  
ESTs cluster 3  
APECED

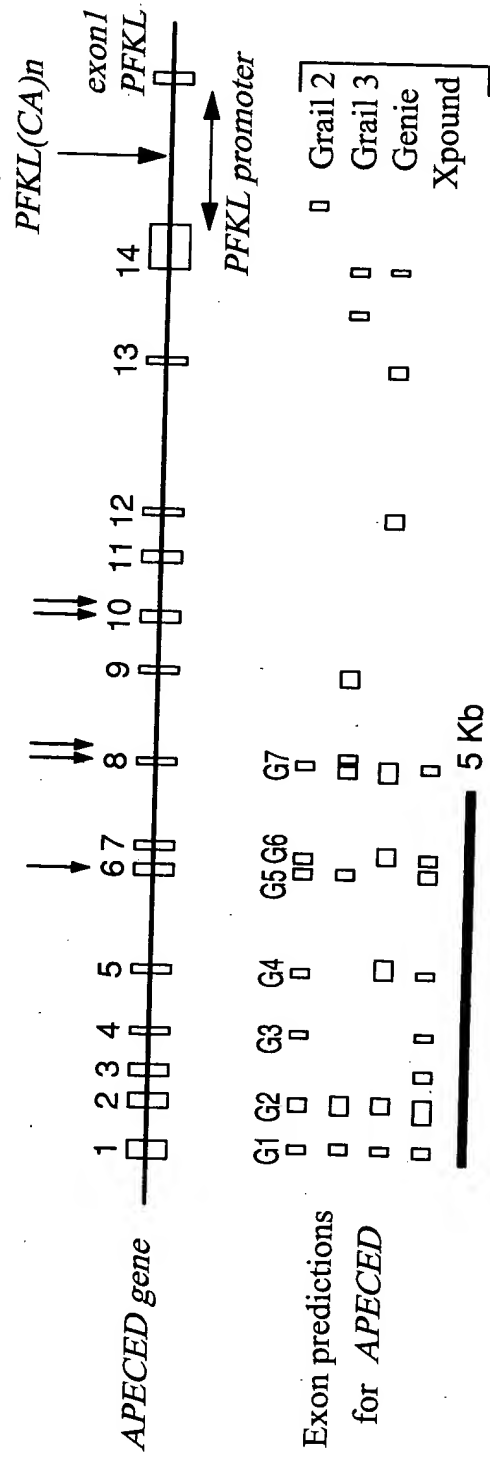


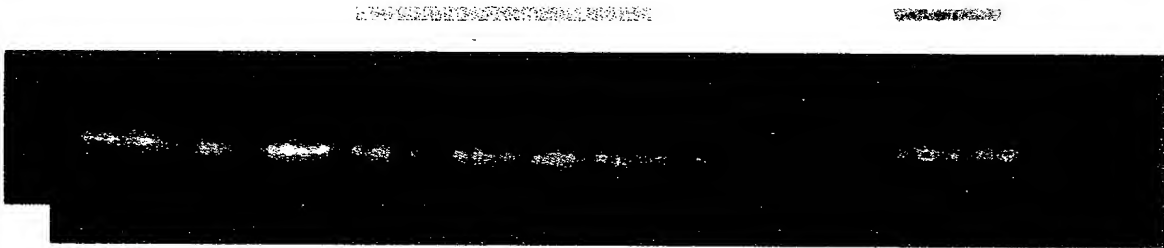
FIG. 1B

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Q11D11

Q21D1

cDNA B1-1



**FIG. 1C**

```

1  cgggcgcacagccggcgaggagccccacagccccgcgggacccgaggccaagcgaggg 60
61  gctgccagtgctcccggaaccacccgcgtccgcgccagccccgggtccccgcgccacccc 120
121 atggcgacggacgcggcgctacgcggcttctgaggtgtcacccgcacggagatcgcgggtg 180
1  M A T D A A L R R L L R L H R T E I A V 20
181 gccgtggacagcgccttcccactgtgtgcacgcgtggctgaccacgacgtggtcccccgag 240
21  A V D S A F P L L H A L A D H D V V P E 40
241 gacaagtcttcaggagacgccttcatctgaaggaaaaaggagggtgtccccaggccttccac 300
41  D K F Q E T L H L K E K E G C P Q A F H 60
301 gccctcctgtcctggctgacctgacccaggactccacagccatccttgacttctggagggtg 360
61  A L L S W L L T Q D S T A I L D F W R V 80
361 ctgttcaaggactacaacctggagcgctatggccggctgcagccatccttgacagcttc 420
81  L F K D Y N L E R Y G R L Q P I L D S F 100
421 cccaaagatgtggacctcagccagccccgggaaggagggaagccccggcgtcccccaag 480
101 P K D V D L S Q P R K G R K P P A V P K 120
481 gctttgggtaccgccaccagactccccaccaagagggaaggcctcagaagggtcgcagct 540
121 A L V P P P R L P T K R K A S E E A R A 140
541 gccgcgcagcagccctgactccaaggggcaccgccagccaggctctcactgaaggcc 600
141 A A P A A L T P R G T A S P G S Q L K A 160
601 aagcccccaagaagcgggagagcagcgagcagcagcgccttccactcgggaacggg 660
161 K P P K K P E S S A E Q Q R L P L G N G 180
661 attcagaccatgtcagcttcagttccagagagctgtggccatgtctcctccgggacgtccccg 720
181 I Q T M S A S V Q R A V A M S S G D V P 200

```

FIG. 2A-1

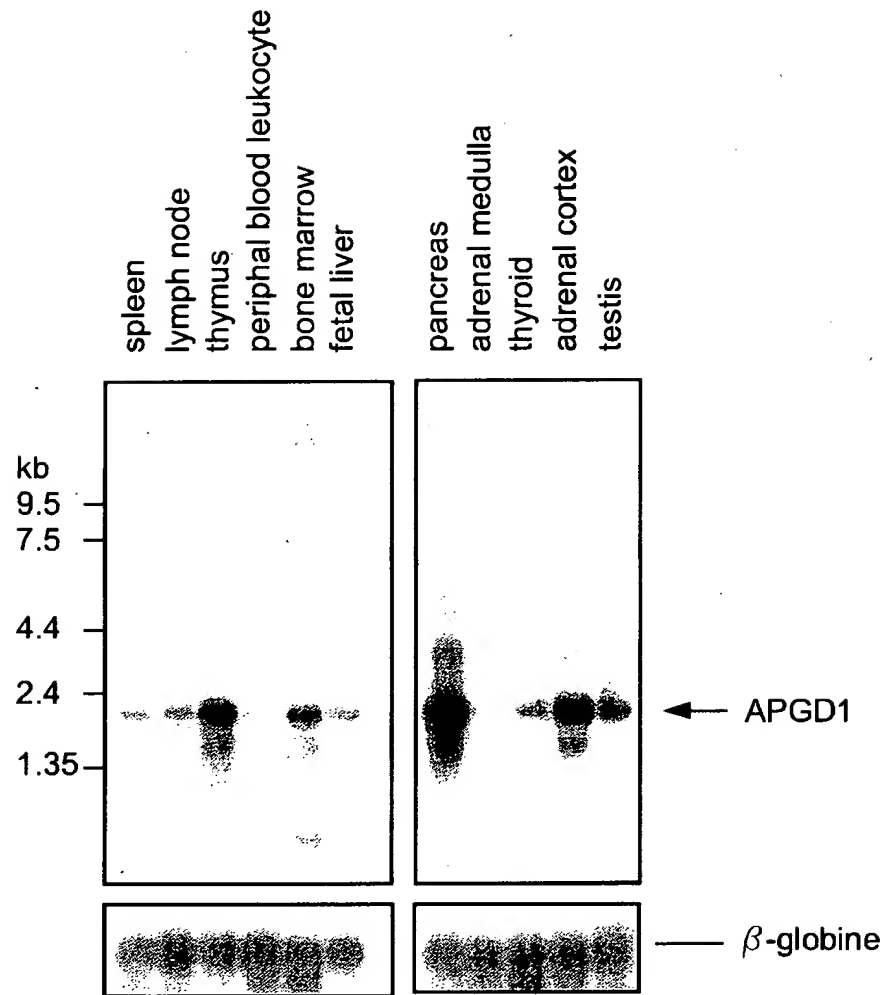
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721	ggagcccgagggccgtggaggggatccctcatccagcagggtgtttgagtcaggcggctcc	780
201	G A R G A V E G I L I Q Q V F E S G G S	220
781	aagaagtgcattccaggttgggtggggagttctacactccagcaagtctcgaagactccggc	840
221	K K C I Q V G G E F Y T P S K F E D S G	240
841	agtgggaagaacaaggcccgagcagcagtgggccgaagcctctgtgttcgagccaagga	900
241	S G K N K A R S S S G P K P L V R A K G	260
901	gcccaggcgctgcccccggtggaggtaggtggccagcagggcagcggttccc	960
261	A Q G A A P G G G E A R L G Q Q G S V P	280
961	gcccccttggccctcccgagtgaccccgagctccaccagaagaatgaggacgagtgtgcc	1020
281	A P L A L P S D P Q L H Q K N E D E C A	300
1021	gtgtgtcgggacggggagctcatctgtgtgacggctgccctcgggccttccacctg	1080
301	V C R D G G E L I C C D G C P R A F H L	320
1081	gcctgcctgtccccctcgctccgggagatccccagtgaggacctggaggtgctccagctgc	1140
321	A C L P P L R E I P S G T W R C S S C	340
1141	ctgcaggcaacagtcaggaggtgcagccccgggcagaggagcccccgccagggagcca	1200
341	L Q A T V Q E V Q P R A E E P R P Q E P	360
1201	cccgtaggagacccccgctcccccggttaggtcggcgaggagaggttaagaggtcca	1260
361	P V E T P L P P G L R S A G E E V R G P	380
1261	cctggggaacccctagcggcatggacacgactcttgtctacaagcacctgcccgtccg	1320
381	P G E P L A G M D T T L V Y K H L P A P	400
1321	ccttctgcagcccccgctgcagggtggactcctcggccctgcacccccctactgtgtg	1380
401	P S A A P L P G L D S S A L H P L L C V	420

FIG. 2A-2

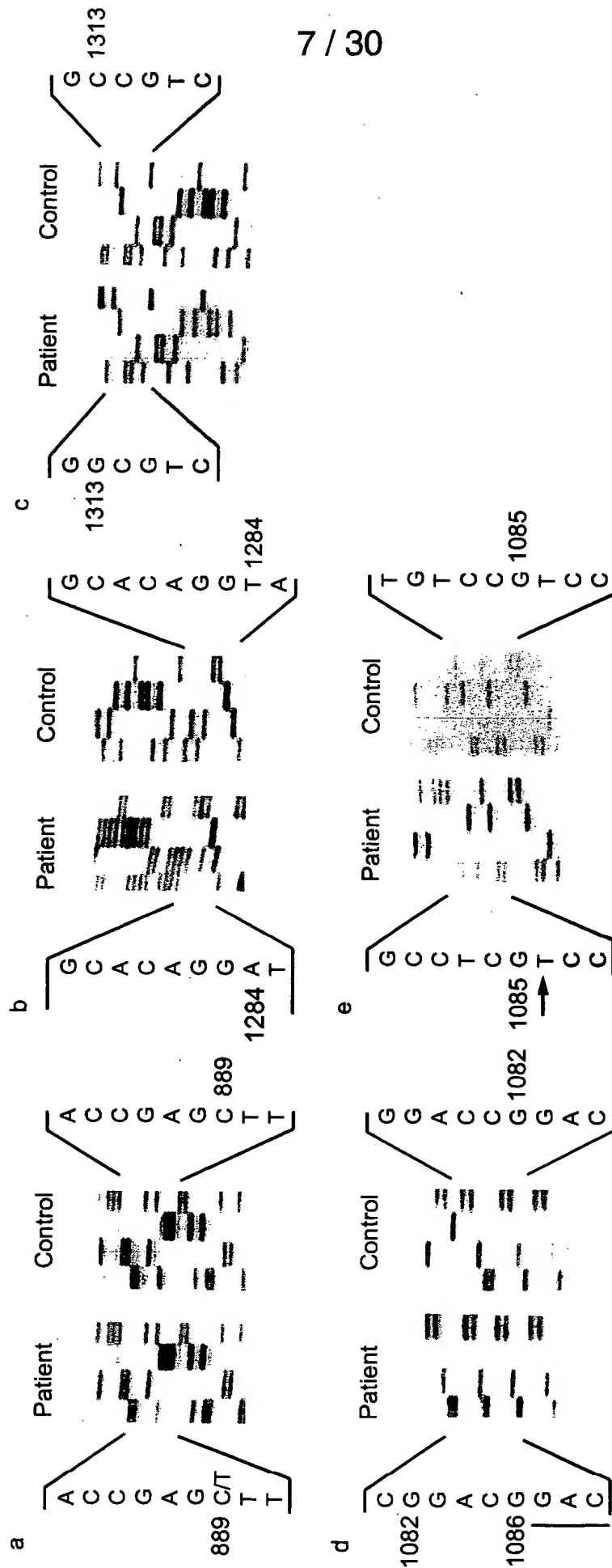
**FIG. 2A-3**

2245



**FIG. 2B**

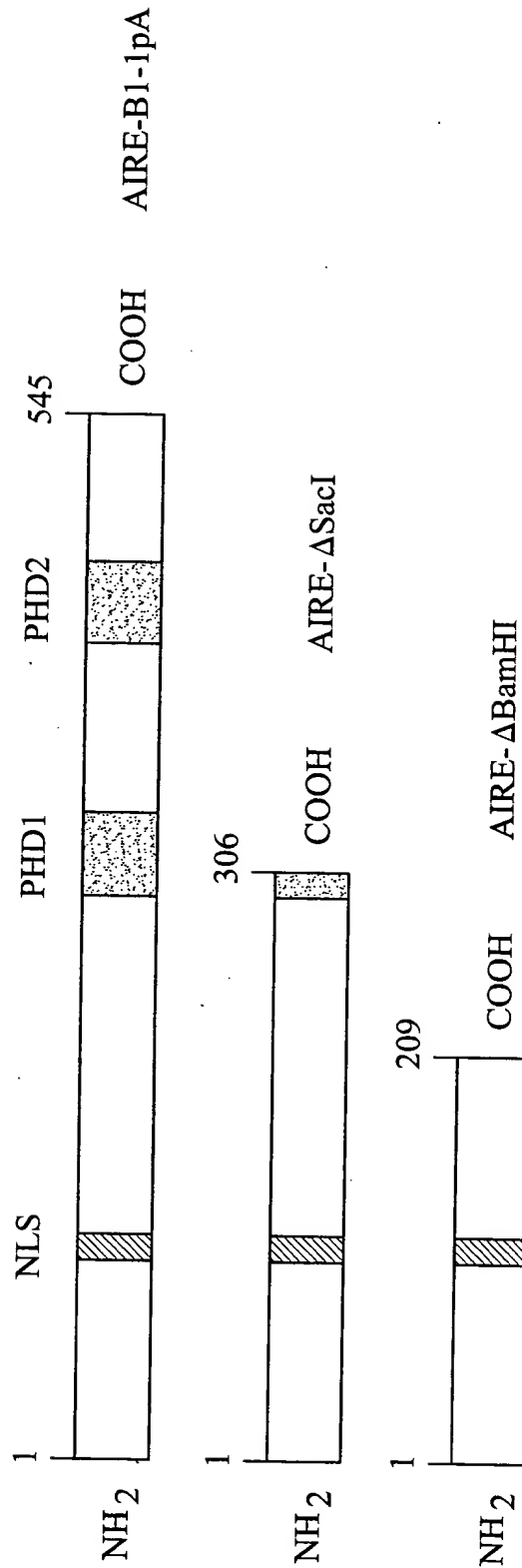
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The sequence lanes appear from left to right, as C, A, T, and G

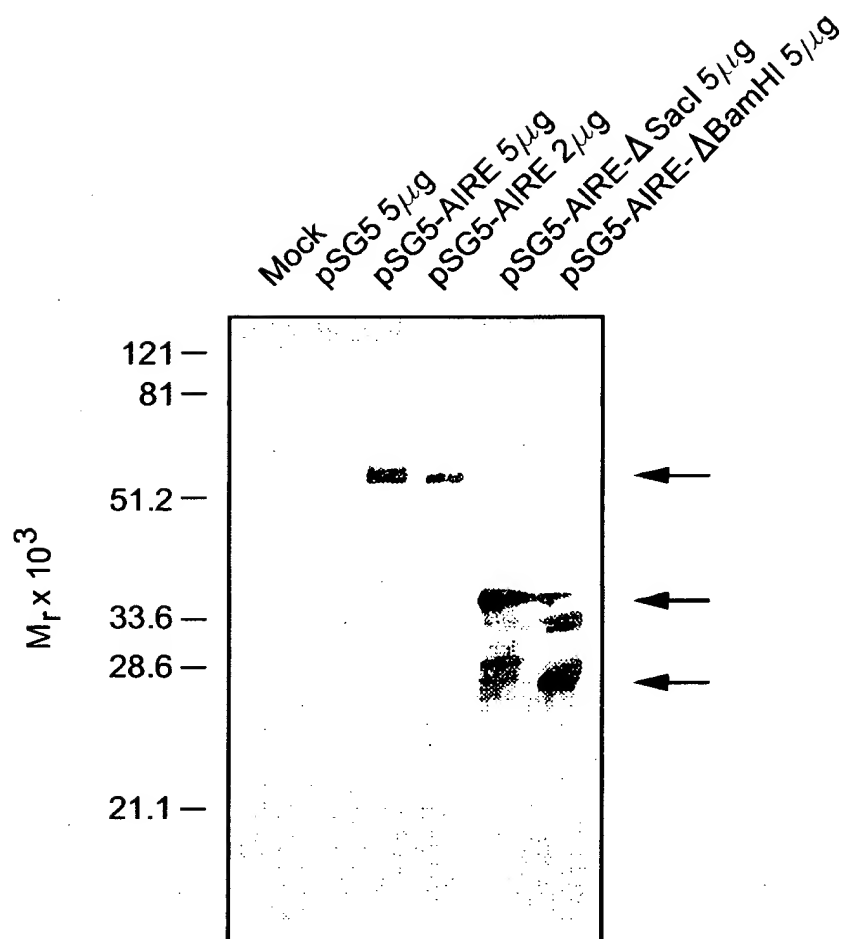
**FIG. 3**

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**FIG. 4**





**FIG. 5**

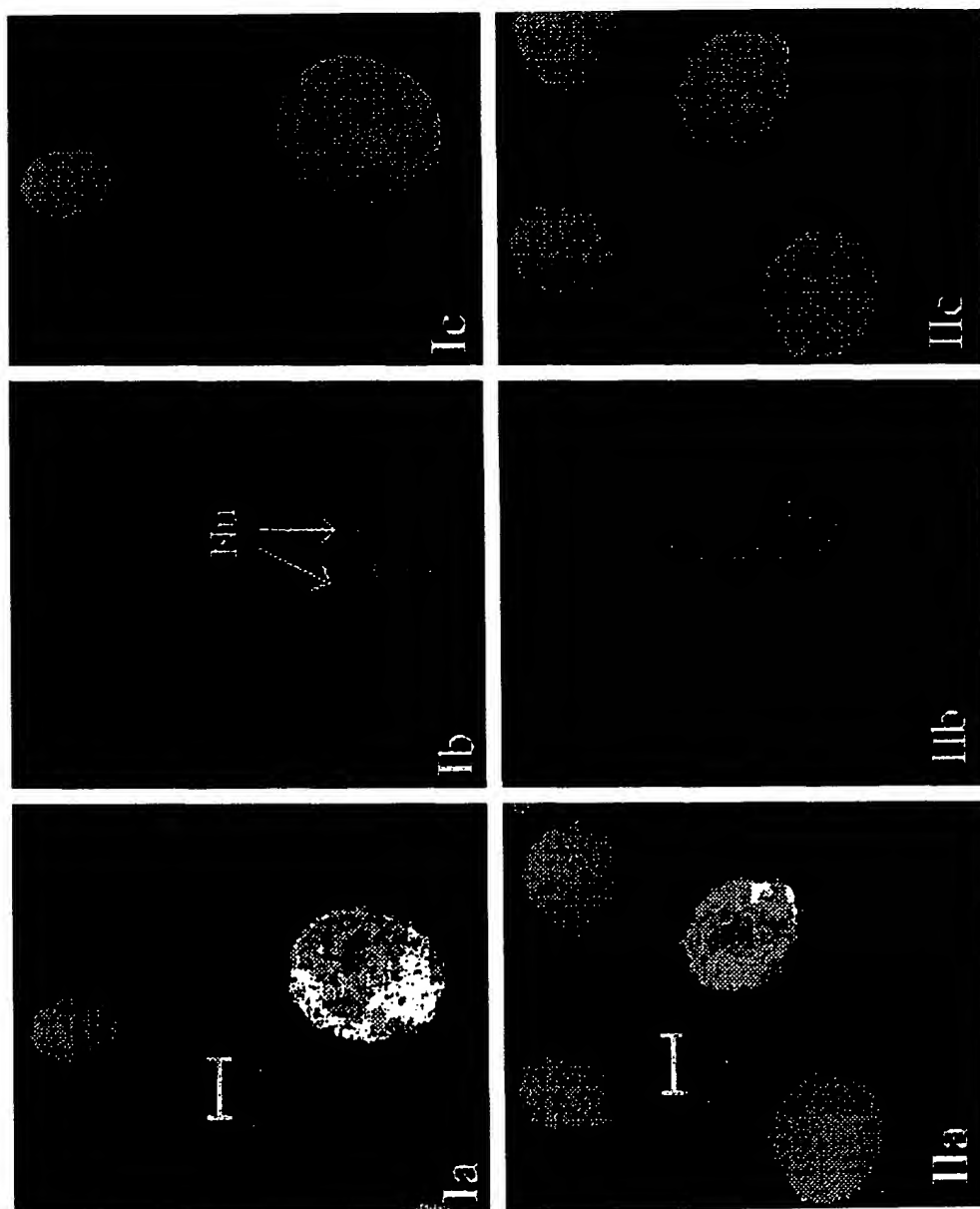


FIG. 6

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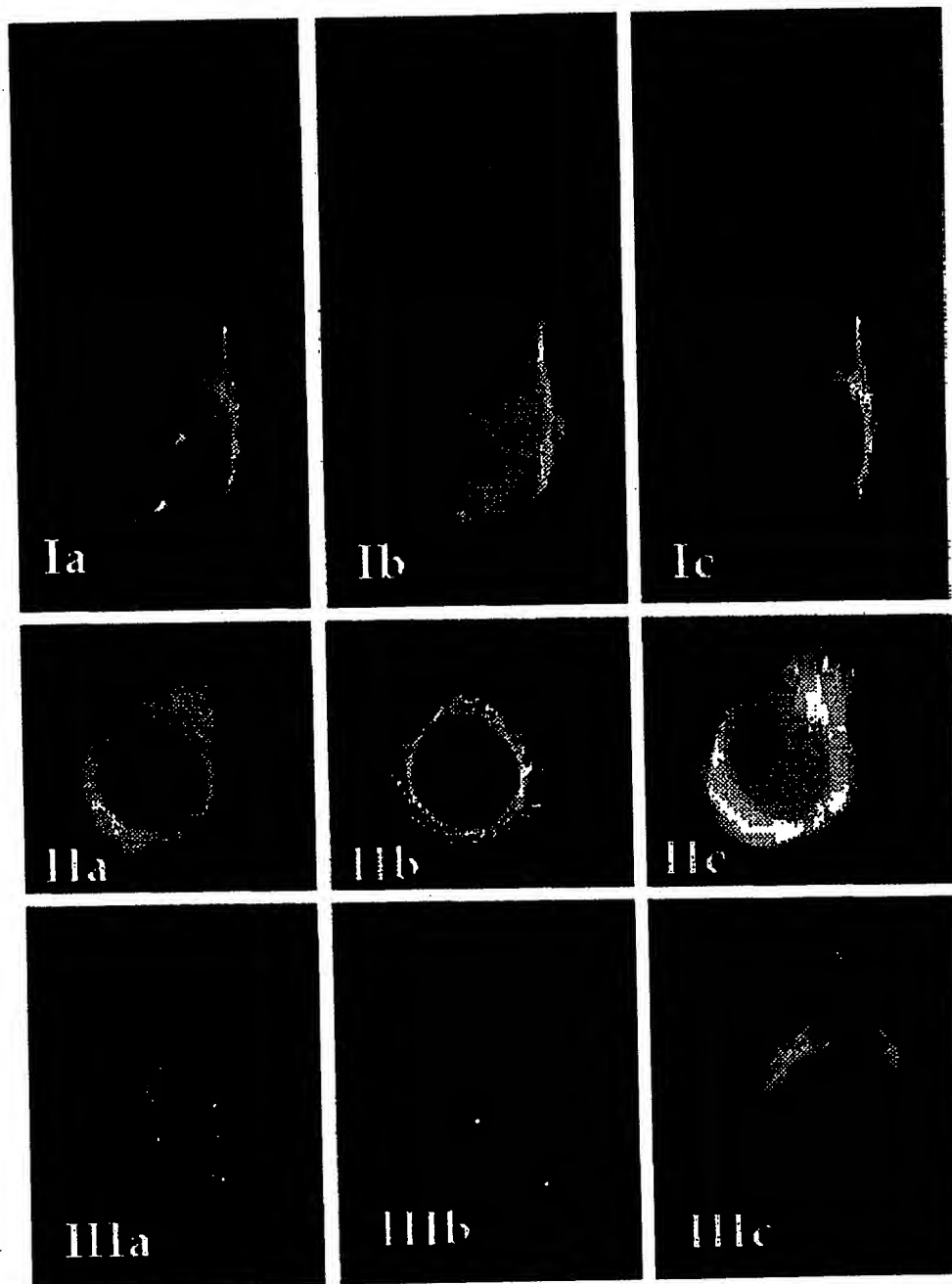
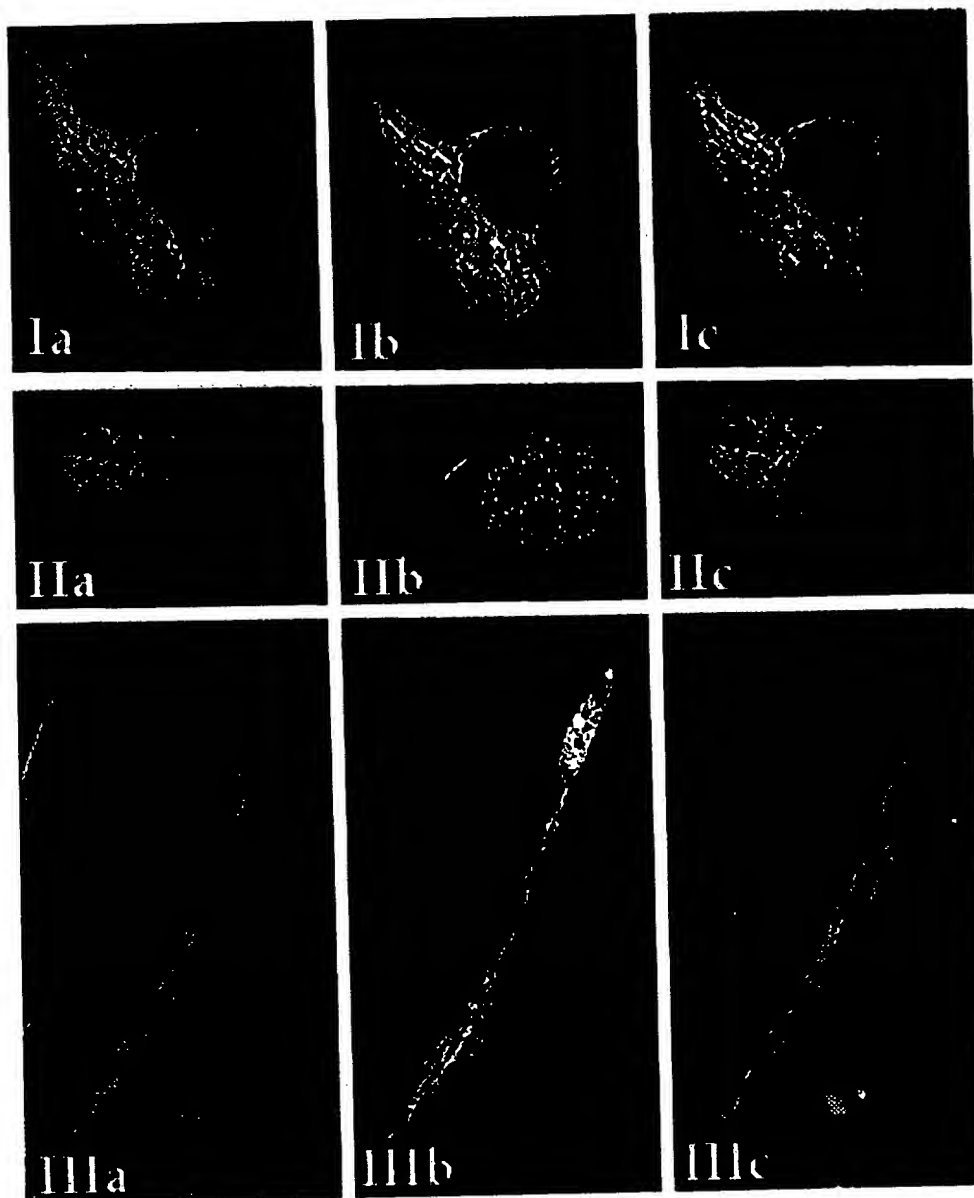
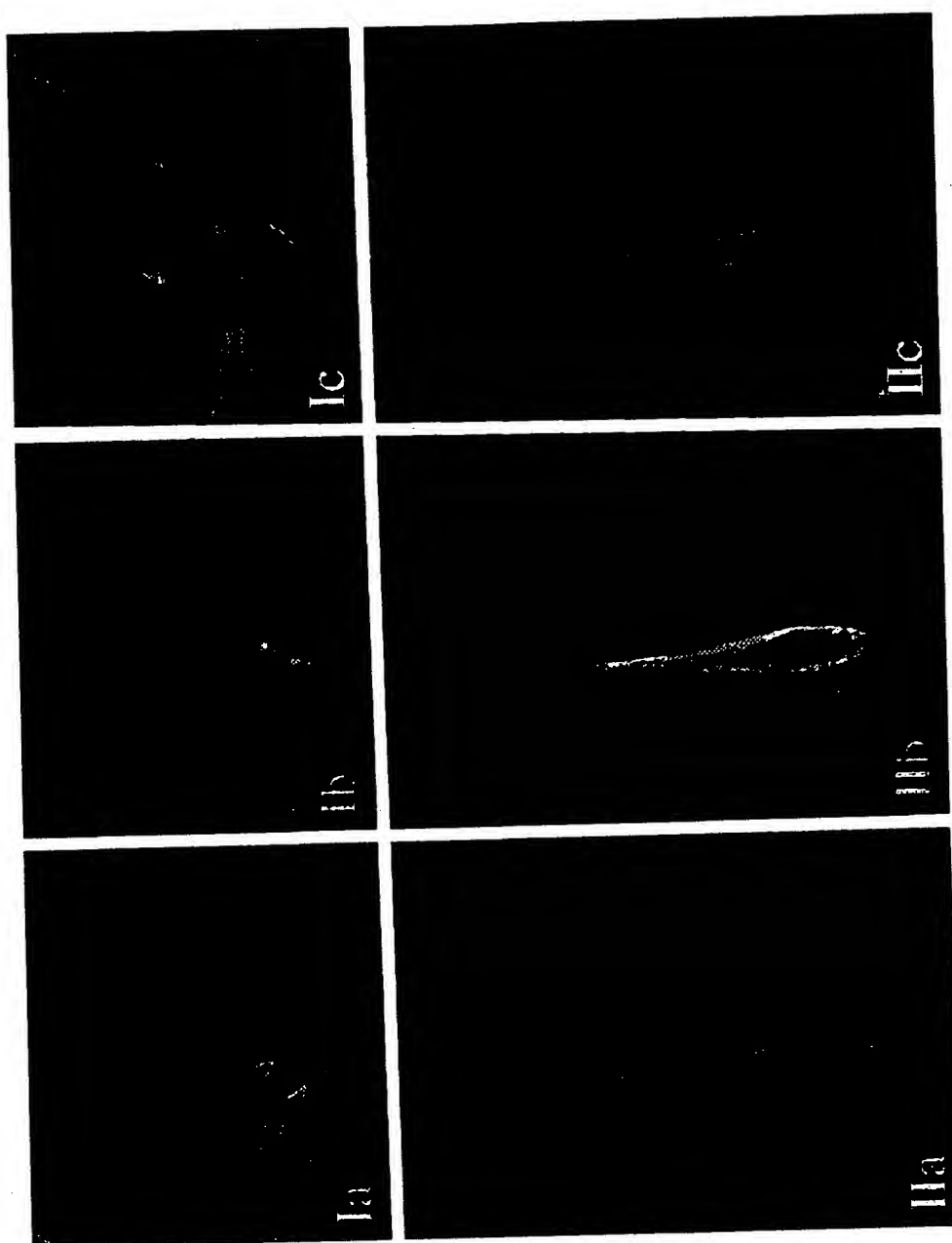


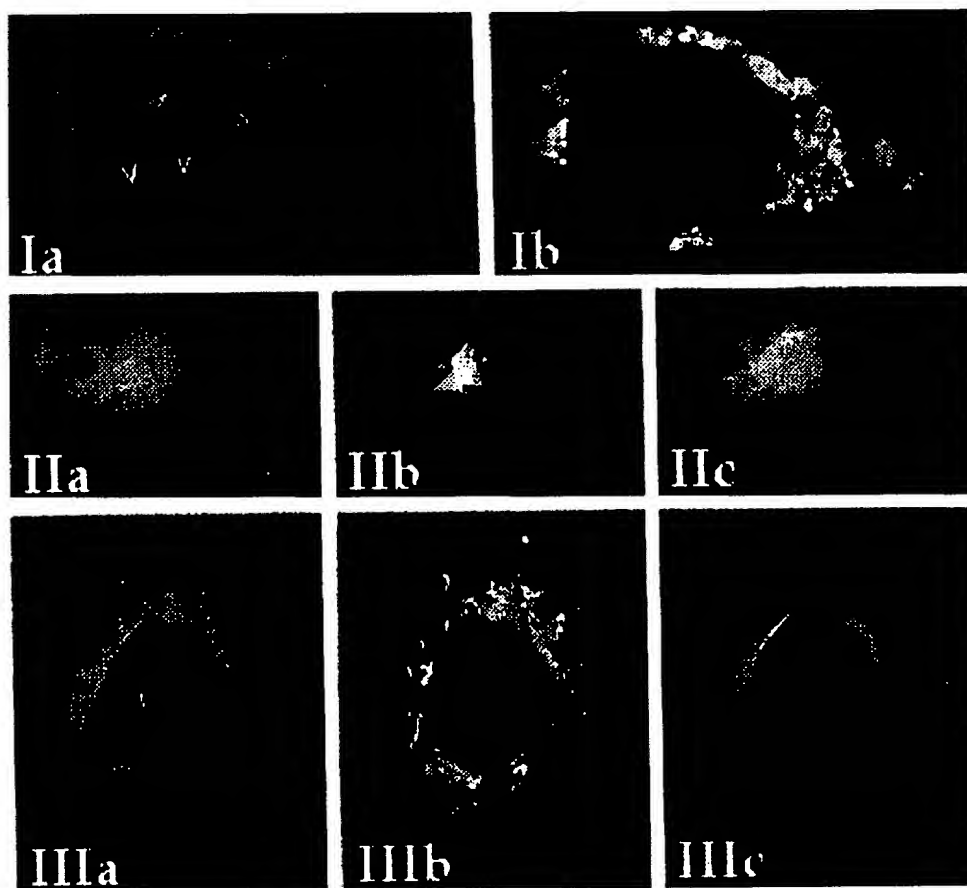
FIG. 7



**FIG. 8**



**FIG. 9**



**FIG. 10**

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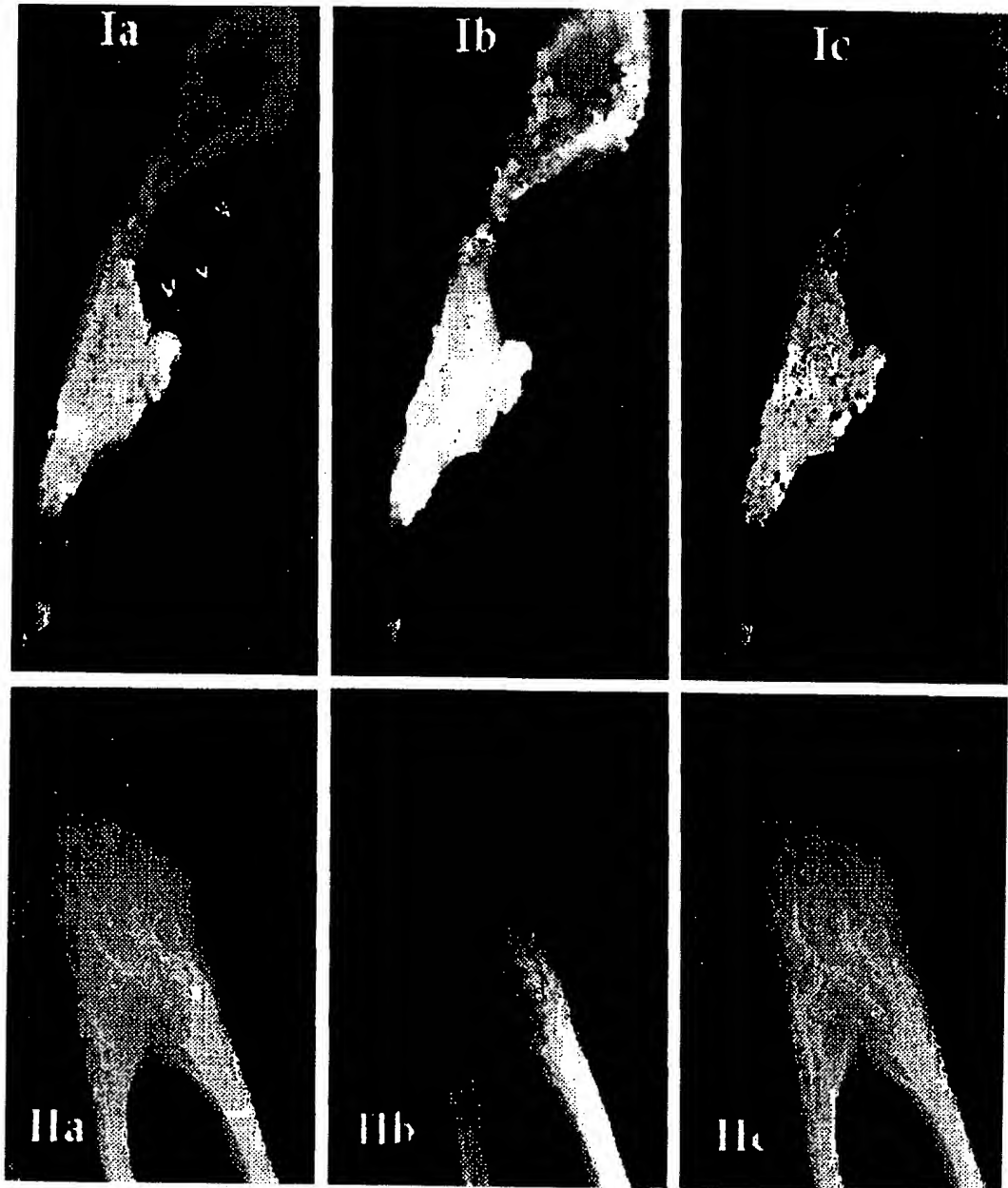


FIG. 11

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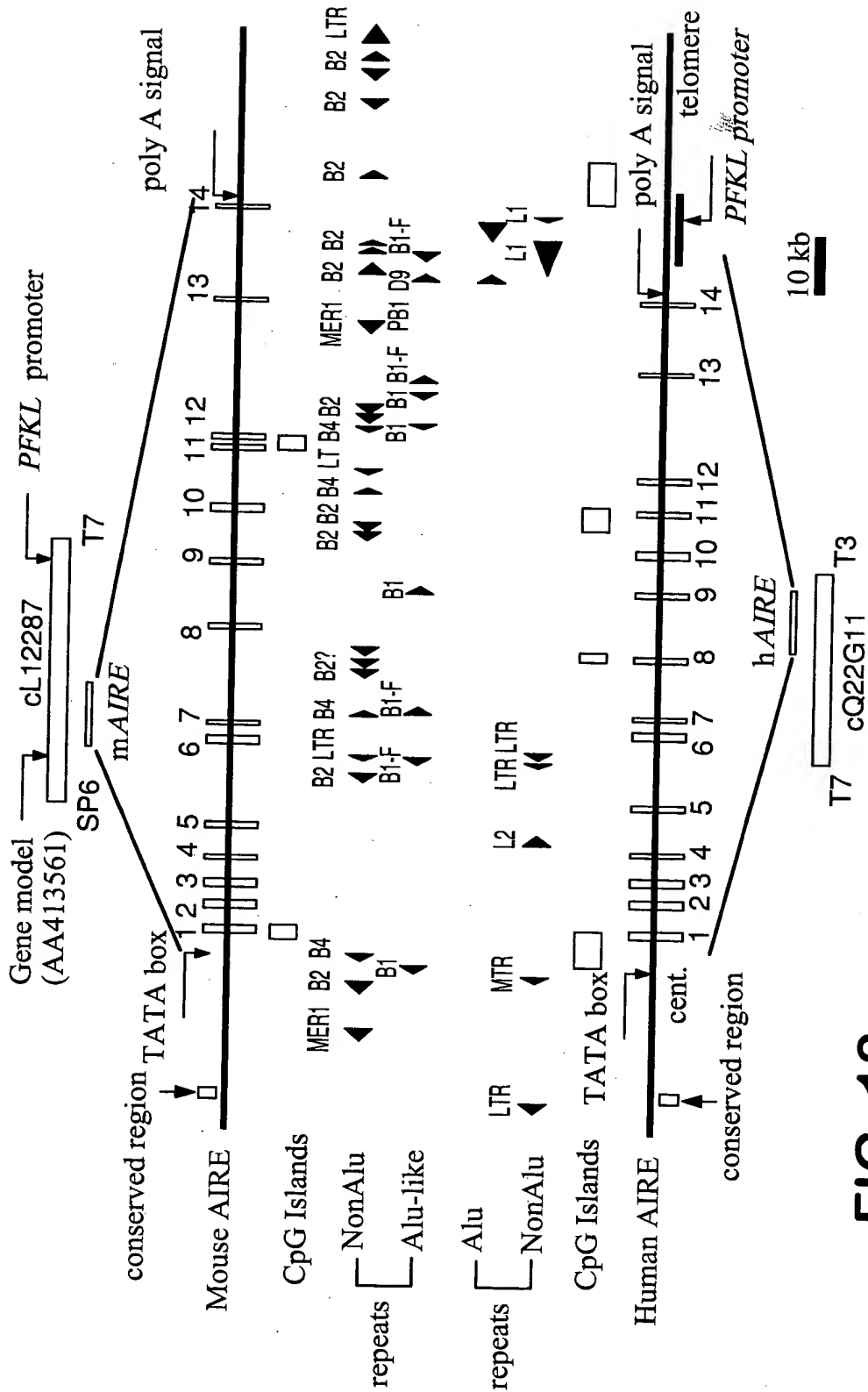


FIG. 12



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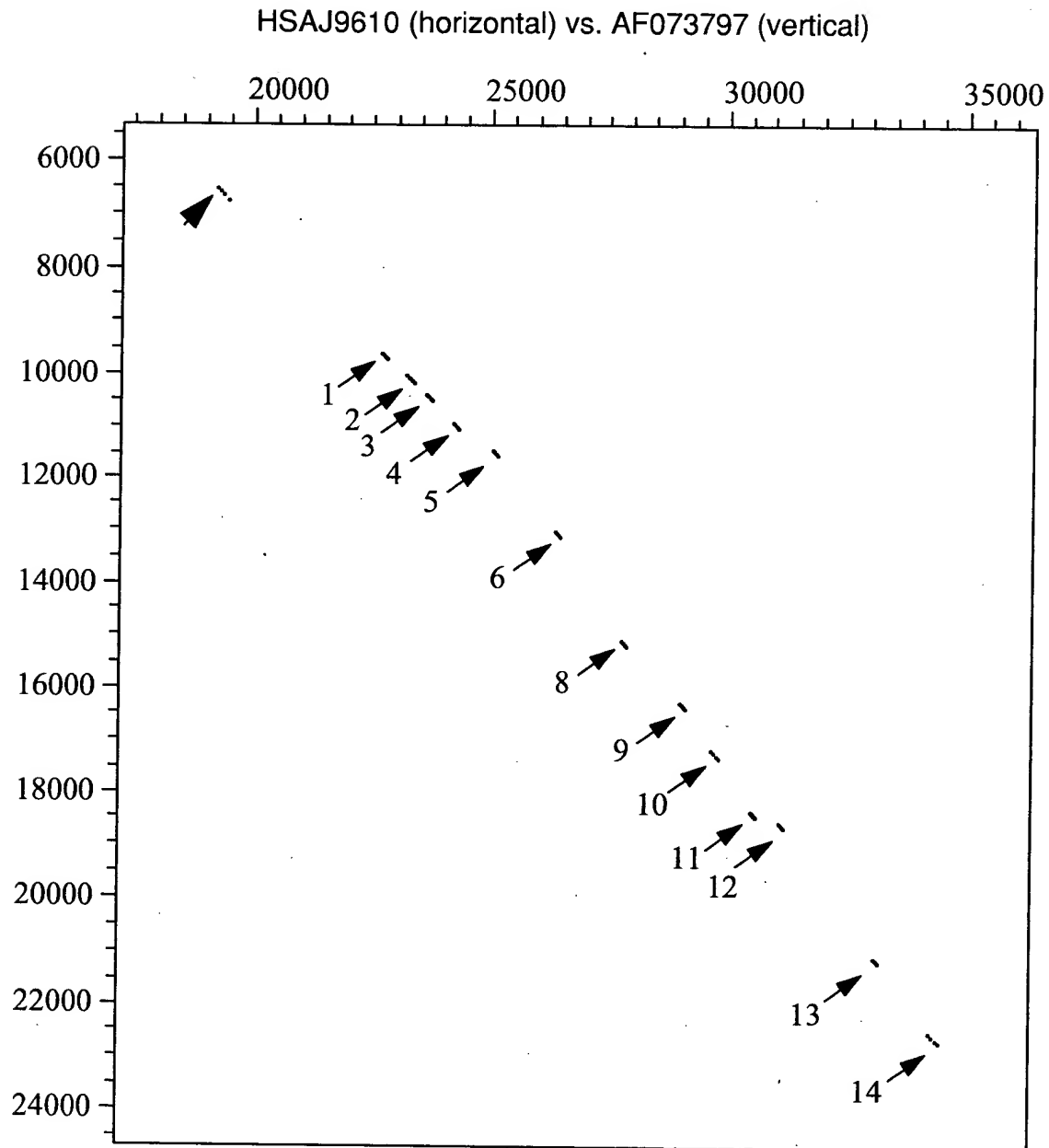


FIG. 13A

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```
6486      GTGTGGACTG TCACGGAAAC CCCACGTGT GATGAAAGT
MAIRE
19186    AAGGGGCTGG TGTGAAAGC CCCACGGCAT GGTGAAAGT
      --G-GG---G T---G-AA-C CCC---G--T G-TGGAAAGT

6575      CCAAAATTCT ACAGGAGTCT TTCTGTTGAT CTCCAGTCAG AGGCTGGGGG
MAIRE
19275    CCGAAATTCT ACAGGGGCCT CTTGTTAAA CCTCCATGCA AGAGGCTGGG
      CC-AAATTCT ACAGG-G-CT -T-TGTT-A- C--C--T--- AG-----GGG
```

FIG. 13B

**FIG. 14-1**

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```

370                               390                               410
361 AAGCGCGGTACTGCCACCCAGACCCCCACCAAGAGAAAGCACTGGAGGCCTCGA 420
-----+-----+-----+-----+-----+-----+-----+
121 K A A V L P P R P P T K R K A L E E P R 140
430                               450                               470
421 GCCACCCCAAGCAACTCTGGCCTCAAAGAGCGTCTCCAGCCCAGGCTCCCACCTGAAG 480
-----+-----+-----+-----+-----+-----+-----+
141 A T P P A T L A S K S V S S P G S H L K 160
490                               510                               530
481 ACTAAGCCCCCTAAGAAGCCAGATGGCAACTTGGAGTCACAGCACCTTCTTGGAAAC 540
-----+-----+-----+-----+-----+-----+-----+
161 T K P P K K P D G N L E S Q H L P L G N 180
550                               570                               590
541 GGAATTCAGACCATGGCAGCTTCTGTCCAGAGAGCTGTGACCGTGGCCTCTGGGATGTT 600
-----+-----+-----+-----+-----+-----+-----+
181 G I Q T M A A S V Q R A V T V A S G D V 200
610                               630                               650
601 CCAGGAACCCGAGGGCCGTGGAAGGATCCTTATCCAGCAGGTGTTGAGTCAGGAAGA 660
-----+-----+-----+-----+-----+-----+-----+
201 P G T R G A V E G I L I Q Q V F E S G R 220
670                               690                               710
661 TCCAAGAAGTGCATTGAGTTGGGGAGAGTTTATACACCCCAAGTTCGAAGACCCC 720
-----+-----+-----+-----+-----+-----+-----+
221 S K K C I Q V G G E F Y T P N K F E D P 240

```

FIG. 14-2

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```

730              750              770
721 AGTGGCAATTGAAACAAGGCCCGAGTGGTAGCAGCCTAAAGCCAGTGGTCCGAGCC 780
-----+-----+-----+
241 S G N L K N K A R S G S S L K P V V R A 260
790      810      830
781 AAGGGAGCCAGGTCACTATACCTGGTAGAGATGAGCAGAAAGTGGGCCAGCAGTGTGGG 840
-----+-----+-----+
261 K G A Q V T I P G R D E Q K V G Q Q C G 280
850      870      890
841 GTTCCTCCCCTTCCATCCCCTCCCCAGTGAGCCCCCAGGTTAACCAAGAAACGAGGATGAG 900
-----+-----+-----+
281 V P P L P S L P S E P Q V N Q K N E D E 300
910      930      950
901 TGTGCCGTGTGCCACGACGAGGTGAGCTCATCTGTTGTGACGGCTGTCCCCGGCCTTC 960
-----+-----+-----+
301 C A V C H D G G E L I C C D G C P R A F 320
970      990      1010
961 CACCTGGCTTGCCTGTCCCCACCTCTGCAGGAGATCCCCAGTGGCCTCTGGAGATGCTCC 1020
-----+-----+-----+
321 H L A C L S P P L Q E I P S G L W R C S 340
1030      1050      1070
1021 TGCTGCCCTCCAGGCAGAGTCCAACAGAAACCTGTCCCAGCCTGAGGTGTCCAGGCCCCCG 1080
-----+-----+-----+
341 C C L Q G R V Q Q N L S Q P E V S R P P 360
-----+-----+-----+

```

FIG. 14-3

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1090	1110	1130	
1081	GAGCTACCTGCAGAGACCCCGATCCTCGTGGGACTGAGGTTCAGATTGAGAAACCAGG		1140
361	E L P A E T P I L V G L R S A S E K T R		380
	1150	1170	1190
1141	GGCCCATCCAGGGAGCTCAAAGCCAGCTCTGATGCTGCTGCATATGTGAACCTGCTG		1200
381	G P S R E L K A S S D A A V T Y V N L L		400
	1210	1230	1250
1201	GCCCCGACCCCTGCAGCTCCTCTGCTGGAGCCCTTCAGCACTGTGCCCTCTACTGAGTGCT		1260
401	A P H P A A P L L E P S A L C P L L S A		420
	1270	1290	1310
1261	GGGAATGAGGGGGCCAGGTCCAGCACCAAGCCGCGATGCAGTGTGTGGCGATGGC		1320
421	G N E G R P G P A P S A R C S V C G D G		440
	1330	1350	1370
1321	ACCGAGGTGTGCGGTGTGCACACTGTGCGCGCTGCCCTTCCACTGGCGCTGCCACTTCCCCG		1380
441	T E V L R C A H C A A A F H W R C H F P		460
	1390	1410	1430

FIG. 14-4

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```
1381 ACGGCCGCGCGCGCGGACCAATCTCCGCTGCAATCCTGCTCTGCAGACTCGACT 1440
-----+-----+-----+-----+-----+-----+-----+-----+
461 T A A A R P G T N L R C K S C S A D S T 480
1450 1470 1490

1441 CCCACGCCAGGCACACCGGGCGAAGCTGTACCCACCTCTGGGCCCCGTCAGCACCTGGG 1500
-----+-----+-----+-----+-----+-----+-----+-----+
481 P T P G T P G E A V P T S G P R P A P G 500
1510 1530 1550

1501 CTTGCCAAGtagGGGACGACTCTGCTAGTCACGACCCCTGTTCTACATAGGGACGACCTG 1560
-----+-----+-----+-----+-----+-----+-----+-----+
501 L A K V G D D S A S H D P V L H R D D L 520
1570 1590 1610

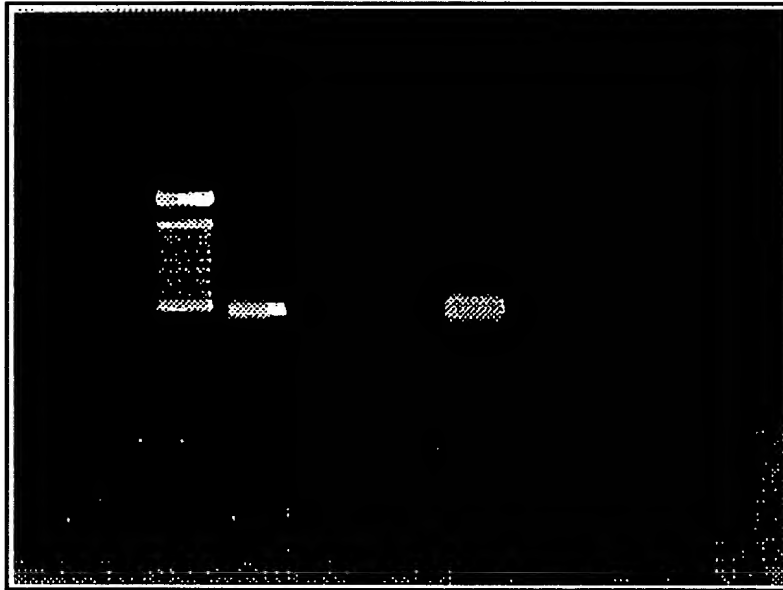
1561 GAGTCCCTCCTCAATGAGCACTCATTTGACGGCATCCTGCAGTGGGCCCATCCAGAGCATG 1620
-----+-----+-----+-----+-----+-----+-----+-----+
521 E S L L N E H S F D G I L Q W A I Q S M 540
1630 1650

1621 TCACGCCCGCTGGCCGAGACACACCCCTTCTCTTCC 1656
-----+-----+-----+-----+-----+-----+-----+-----+
541 S R P L A E T P P F S S 552
```

FIG. 14-5

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M 1 2 3 4 5 6



**FIG. 15**



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Human AIRE	-MATDAFLRR LRLHRTEIA VAVDSAFPLL HALADHDVVP EDKFQETLHL	
Mouse AIRE	MAGGDGMLRR LRLHRTEIA VAIDSAFPLL HALADHDVVP EDKFQETLRL	
Consensus	----D--LRR LRLHRTEIA VA-DSAFPLL HALADHDVVP EDKFQETL-L	
Human AIRE	51 KEKEGCPQAF HALLSWLLTQ DSTAILDFWR VLFKDYNLER YGRLOPILDS	100
Mouse AIRE	KEKEGCPQAF HALLSWLLTR DSGAILDFWR ILFKDYNLER YSRLHSILDG	
Consensus	KEKEGCPQAF HALLSWLLT- DS-AILDFWR -LFKDYNLER Y-RL--ILD-	
Human AIRE	101 FPKDVDLSQP RKGRKPPAVP KALVPPPRLP TKRKASEEAR AAAPAAALTPR	150
Mouse AIRE	FPKDVDLNQS RKGRKPLAGP KAAVLPPRPP TKRKALEEPR ATPPATLASK	
Consensus	FPKDVDL-Q- RKGRKP-A-P KA-V-PPR-P TKRKA-EE-R A--PA-L---	
Human AIRE	151 GTASPGSQLK AKPPKKPESS AEQRLPLGN GIOTMSASVQ RAVAMSSGDV	200
Mouse AIRE	SVSSPGSHLK TKPPKKPDGN LESQHLPLGN GIOTMAASVQ RAVTVASGDV	
Consensus	---SPGS-LK -KPPKKP--- -E-Q-LPLGN GIOTM-ASVQ RAV---SGDV	
Human AIRE	201 PGARGAVEGI LIQQVFESGG SKKCIQVGGE FYTPSKFED. SGSGKNKARS	250
Mouse AIRE	PGTRGAVEGI LIQQVFESGR SKKCIQVGGE FYTPNKFEDP SGNLKNKARS	
Consensus	PG-RGAVEGI LIQQVFESG- SKKCIQVGGE FYTP-KFED- SG--KNKARS	
Human AIRE	251 SSGPKPLVRA KGAQGAAPGG GEARLQQQGS VPAPLALPSD PQLHQKNEDE	300
Mouse AIRE	GSSLKPVVRA KGAQVTIPGR DEQKVGQCG VPPLPSLPSE PQVNQKNEDE	
Consensus	-S--KP-VRA KGAQ---PG- -E---GQQ-- VP----LPS- PQ--QKNEDE	

FIG. 16-1

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Human AIRE	301	CAVCRDGGEL	ICCDGCPRAF	HLACLSPPLR	EIPSGTWRC	SCLQATVQEV	350
Mouse AIRE		CAVCHDGGEL	ICCDGCPRAF	HLACLSPPLO	EIPSGLWRC	CCLQGRVQQN	
Consensus		CAVC-DGGEL	ICCDGCPRAF	HLACLSPPL-	EIPSG-WRC	-CLQ--VQ--	
Human AIRE	351	QPRAEPRPQ	EPVETPLPP	GLRSAGEVR	GPFGEPLAGM	DTTLVYKHL	400
Mouse AIRE		LSQPEVSRPP	ELPAETPILV	GLRSASEKTR	GPSRELKASS	DAAVTYVNL	
Consensus		----E--RP-	E-P-ETP---	GLRSA-E--R	GP--E--A--	D-----Y--L-	
Human AIRE	401	APPSAAPLPG	LDSSALHPLL	CVGPEGQONL	APGARCGVCG	DGTDVLRCTH	450
Mouse AIRE		APHPAAPL..	LEPSALCPLL	SAGNEGRPGP	APSARCSVCG	DGTEVLRCAH	
Consensus		AP--AAPL--	L--SAL-PLL	--G-EG----	AP-ARC-VCG	DGT-VLRC-H	
Human AIRE	451	CAAAFHWRCH	FPAGTSRPGT	GLRCRSCSGD	VTPAP.VEGV	LAP.SPARLA	500
Mouse AIRE		CAAAFHWRCH	FPTAAARPOT	NLRCKSCSAD	STPTPGTPE	AVPTSGPRPA	
Consensus		CAAAFHWRCH	FP----RPOT	-LRC-SCS-D	-TP-P---G-	--P-S--R-A	
Human AIRE	501	PGPAK...DDT	ASHEPALHRD	DLESLLSEHT	FDGILQWAIQ	SMARPAAPFP	550
Mouse AIRE		PGLAKVGDD	ASHDFVLHRD	DLESLLNEHS	FDGILQWAIQ	SMSRPLAETP	
Consensus		PG-AK--DD-	ASH-P-LHRD	DLESLL-EH-	FDGILQWAIQ	SM-RP-A--P	
Human AIRE	551	S---					
Mouse AIRE		PFSS					
Consensus		----					

FIG. 16-2

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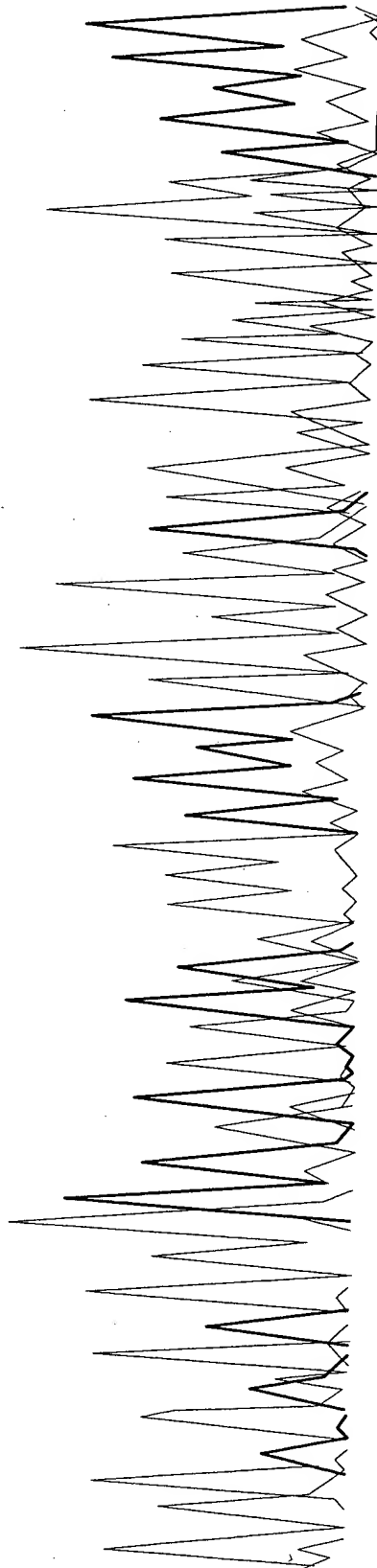
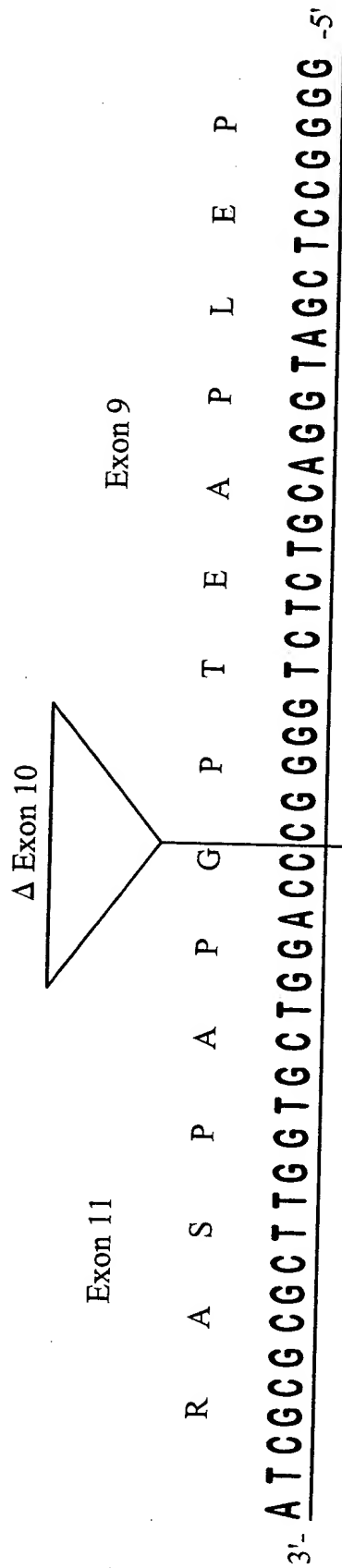


FIG. 17A

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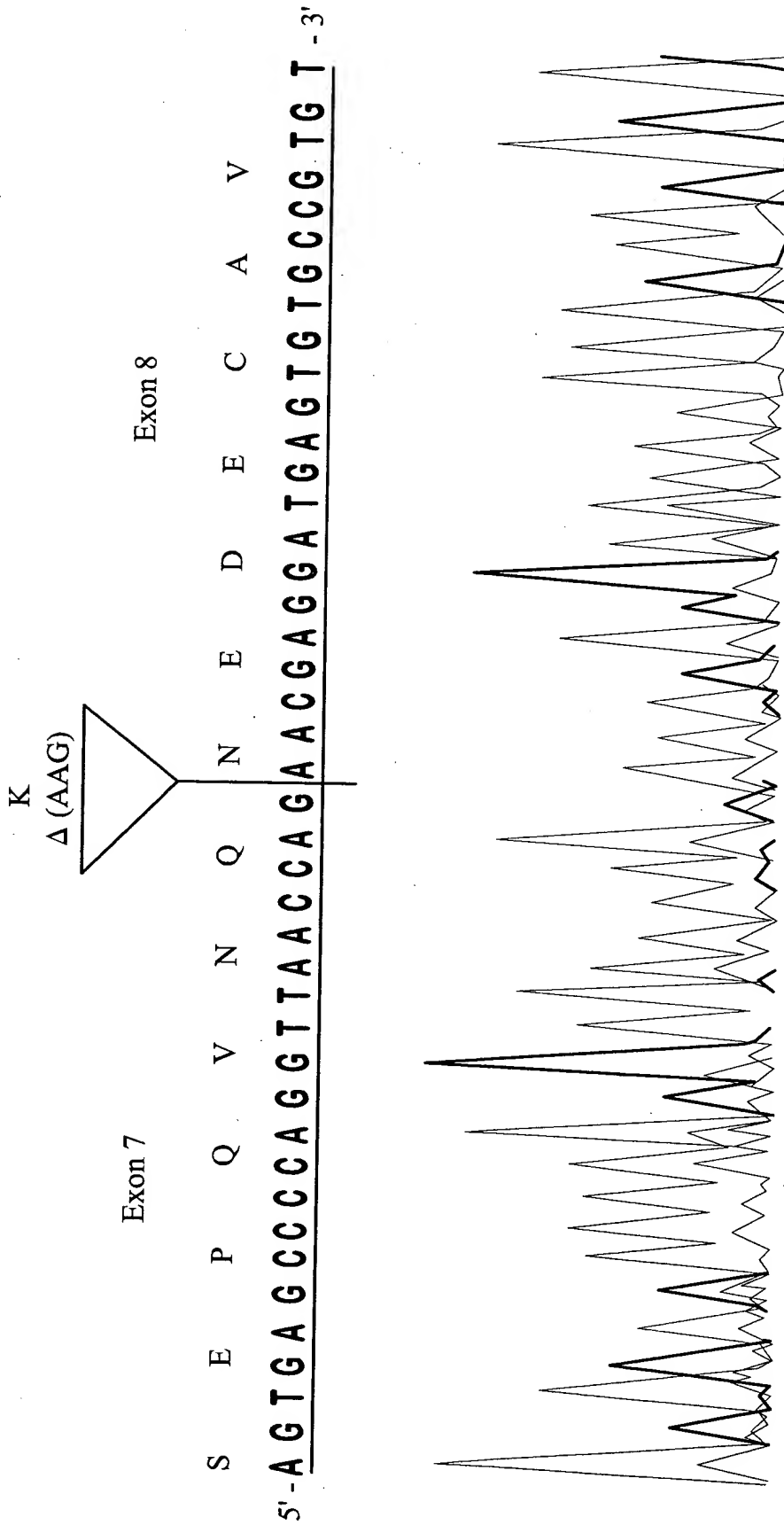


FIG. 17B

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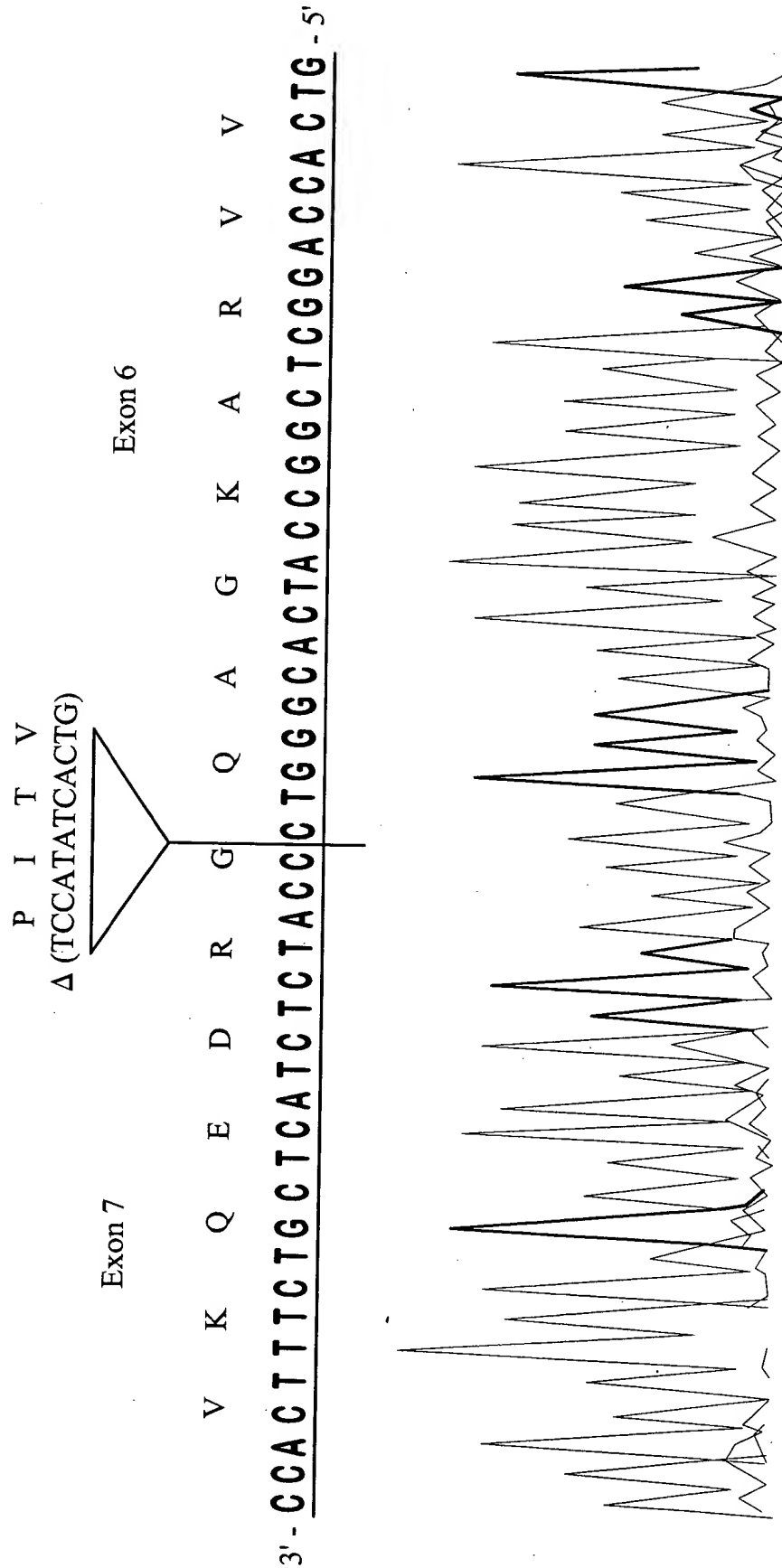
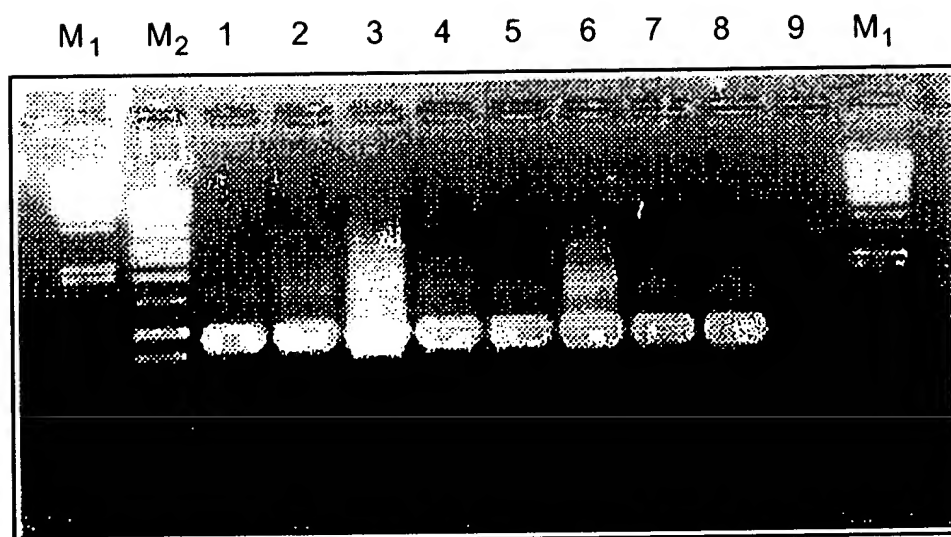


FIG. 17C



**FIG. 18**

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